TÜSEB TAÇESE
TÜRKİYE MATERNITY, CHILD AND ADOLESCENT HEALTH INSTITUTE

Duchenne Muscular Dystrophy (DMD) Experiences and Recommendations from the Perspective of Patients and Families

WORKSHOP REPORT

September 17, 2025

Online

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Message from the President

The Health Institutes of Türkiye (TÜSEB) pioneers innovative, systematically conducted studies in the field of health sciences and technologies, providing scientific, technical, and financial support to practitioners and researchers. In this context, it operates with the mission of fostering a domestic and national healthcare sector in Türkiye, alongside the vision of becoming a leading international R&D-focused organization. Under the motto "Science and Technology for Human Health", it aims to bring new momentum and energy to the healthcare sector. To enhance and sustain Türkiye's competitiveness, it supports all kinds of scientific and technological endeavors that hold the potential to add value to our country and humanity.

The Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE) is one of the nine institutes established under TÜSEB. With the motto "Healthy Mother, Healthy Child, Healthy Generations", the objectives of our Institute include: "To ensure the production, application, and utilization of techniques and methods aimed at identifying causes, preventing, diagnosing, and treating diseases related to maternal, child, and adolescent health; to establish collaborations and partnerships with relevant institutions and organizations in the private, foundation, or public sectors for this purpose; and to support entrepreneurship in these fields."

The childhood and adolescence periods, which fall within our scope of activity, are known as the stages where individuals experience the most rapid physical, mental, and social development. Adequate healthcare services and supportive environments provided at an early age help children and youth maximize their developmental potential. In this context, to achieve a healthy society, it is essential to develop and implement appropriate health policies tailored to these specific life stages.

We have organized the DMD awareness meeting titled "Experiences and Proposed Solutions from the Patient's Perspective" with the aims of evaluating Duchenne Muscular Dystrophy (DMD), a rare childhood disease, from the perspective of patients and families, identifying needs, and shaping recommendations for projects and policies to be developed. The meeting was held with the participation of our Institute, academicians, experts, representatives of relevant non-governmental organizations representing DMD patients and their families, and families struggling with the disease. In this meeting, which is anticipated to guide science and technology studies aimed at protecting and improving the health of patients and families, as well as future workshops, participants were informed about the institution, current debates regarding DMD were discussed, and the experiences of DMD patients and families regarding the disease and care processes, their health needs, and proposed solutions for these needs were evaluated. The views and suggestions presented during the meeting have been systematically reported and submitted for the evaluation of the relevant parties.

Hoping that the outcomes will be beneficial to our country...

Prof. Dr. Şirin Güven

President of the Türkiye Maternity, Child and Adolescent Health Institute

Executive Summary

Due to its progressive course, genetic nature, and high care burden, Duchenne Muscular Dystrophy (DMD) is not merely a medical condition; it is also a multidimensional issue of health, social security, and education policy. The "DMD Awareness Meeting: Experiences and Proposed Solutions from the Patient's Perspective", held on September 17, 2025, under the coordination of the Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE), aimed to reveal the unmet needs of individuals and families struggling with this challenging disease through evidence based on the realities of the field.

Data obtained from this large-scale meeting with 70 participants, comprising public authorities, academicians, specialist physicians, non-governmental organizations, and patients' relatives, demonstrated that fairness in access to health services, alleviating the caregiver burden, and removing bureaucratic obstacles are just as critical in DMD management as clinical research.

Participating relatives of DMD patients and representatives of DMD associations expressed their experiences regarding access to health services, medical devices, and treatments, along with their views and suggestions. Accordingly, the prominent expectations and policy recommendations regarding DMD disease are as follows:

- **Multidisciplinary Centers:** Establishing centers in university and city hospitals that bring together relevant disciplines and provide DMD-specific diagnosis, treatment, and rehabilitation services, and strengthening existing centers.
- **Home Care and Telehealth Services:** Strengthening home care services, providing psychological counseling support, enabling ventilator and orthosis measurements to be done at home, and delivering technical team support to families.
- **Health Report Processes:** Making long-term report arrangements taking into account the chronic and progressive course of the disease, since the obligation of annual report renewal creates a serious burden on families.
- **Psychosocial Support Programs:** Establishing regular psychological and social support mechanisms to protect the mental health of patients (especially adolescent children with DMD) and families under a heavy care burden.
- **Education and Social Participation:** Conducting efforts to remove the obstacles experienced in the processes of school enrollment and continuation of education for children with DMD in practice, despite existing legislative regulations, and ensuring equal opportunities in education.
- **Access to Medical Devices:** Bringing vital cough assist devices, high-quality wheelchairs, and orthoses under full SSI (Social Security Institution) coverage; and delivering them directly to homes through a "device prescription model".
- **Domestic R&D and Drug Development Strategies:** Supporting domestic drug and treatment research conducted in Türkiye, allocating special funds for rare diseases, and accelerating

processes.

- **Preventive Health Policies and Genetic Counseling:** Making premarital genetic screenings a routine public health policy and providing priority genetic counseling to risk groups with a family history.
- **Protection Against Unethical Practices:** Protecting families against scientifically invalid applications characterized as false hope peddling, and developing preventive regulations by health authorities.

In conclusion; this report is not merely a situation assessment, but a fundamental needs analysis based on the experiences of patients and families. It is anticipated that the prepared report will contribute as a guiding document for relevant institutions and decision-makers and form a strong foundation for policy development processes.

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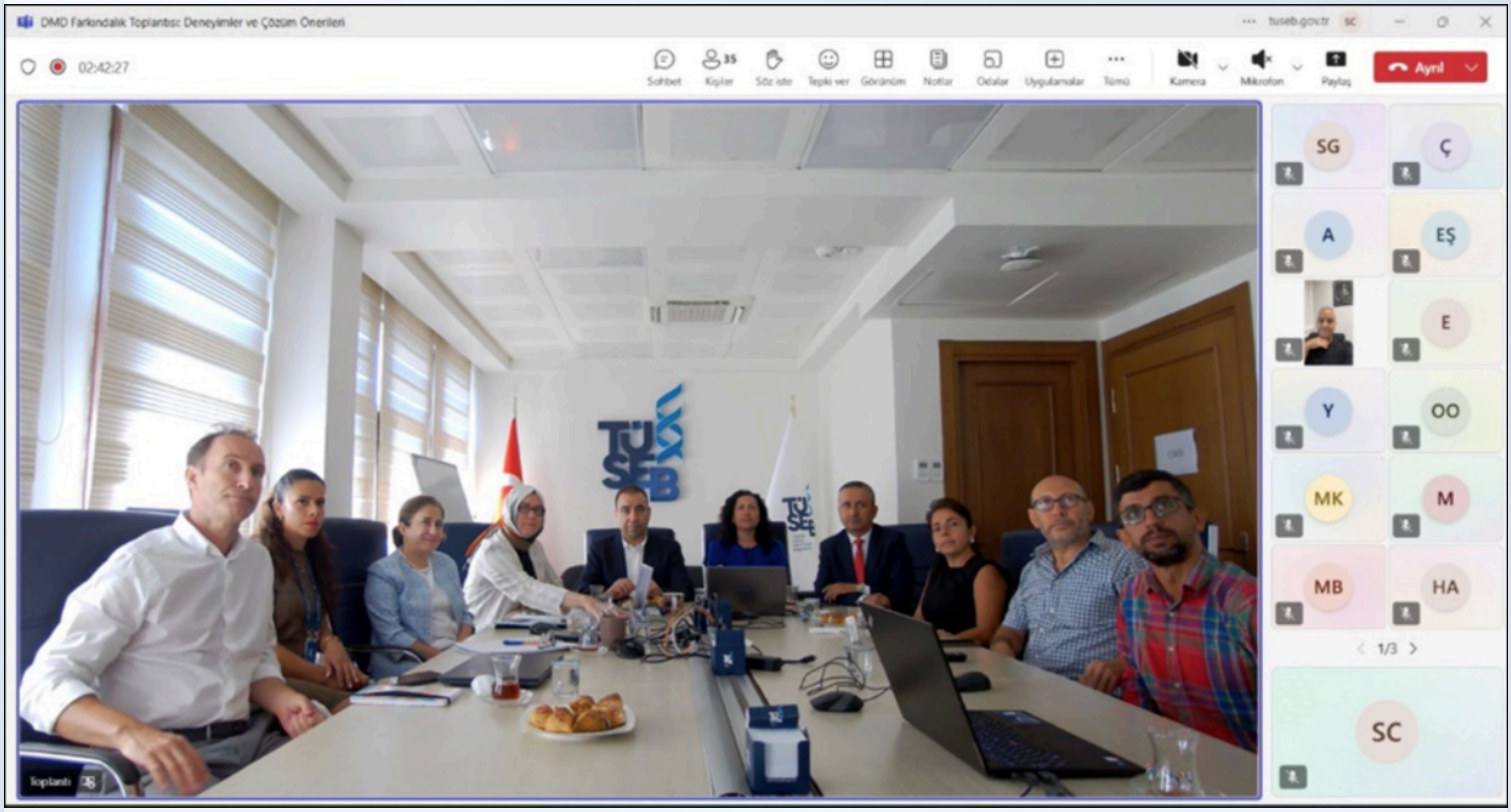

Duchenne Muscular Dystrophy (DMD) Experiences and Recommendations from the Perspective of Patients and Families

Online

SEPTEMBER 17, 2025 • ONLİNE

1. TAÇESE DMD Awareness Meeting: Experiences and Proposed Solutions from the Patient's Perspective

Duchenne Muscular Dystrophy (DMD) holds a unique and critical position within the spectrum of rare diseases due to both its incidence rate and the multidimensional impacts it causes. Although classified as "rare", occurring in approximately one out of every 3,500–5,000 live male births worldwide, DMD is one of the most common pediatric neuromuscular diseases and generates one of the highest care burdens. The progressive course of the disease and its lifelong effects transform DMD from being merely a clinical condition into a health priority that requires comprehensive and holistic approaches for health systems.

One of the distinguishing features of DMD within the rare disease ecosystem is the prominence of its multidisciplinary care requirement. The disease process necessitates the coordination of neurology primarily, as well as cardiology, pulmonology, endocrinology, physical therapy and rehabilitation, and psychosocial support services. In this aspect, DMD represents one of the complex and challenging application areas of the "holistic care" approach in rare disease management. On the other hand, due to genetic treatments and orphan drug development processes aimed at the DMD disease, it concurrently comes to the fore alongside biotechnological innovation, regulatory frameworks, and ethical evaluations.

Beyond clinical management, DMD is also important in terms of preventive health services and public health strategies due to its genetic transmission. The hereditary nature of the disease highlights interventions such as the development of premarital screening programs, the provision of comprehensive genetic counseling services for at-risk families, and making prenatal diagnostic opportunities accessible within the scope of prevention principles.

Within this multilayered structure, effective and sustainable policy development processes in the field of DMD cannot be limited solely to clinical research. In lifelong diseases that require a high care burden, feedback based on the direct experiences of patients and families stands out as a complementary component of evidence-based policy making. Identifying unmet needs, making the caregiver burden visible, evaluating the accessibility of health and social services, and ensuring fairness and transparency in access to treatment are only possible by incorporating the patient and family perspective into the process.

Within this framework, the DMD Awareness Meeting held on September 17, 2025, under the coordination of the Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE), aimed to systematically address the experiences, common problem areas, and proposed solutions of patients and families in the field of Duchenne Muscular Dystrophy. The views shared during the meeting were evaluated under thematic headings, isolated from individual narratives; it was targeted to provide input for policy and project studies that can be developed in the areas of

healthcare delivery, multidisciplinary care models, rehabilitation, psychosocial support, education, and research.

This report was prepared by compiling the findings and recommendations put forward during the meeting process based on the patient and family perspective, and it is anticipated to serve as a reference document for practices, scientific research, and policy development activities to be conducted in the field of rare diseases.

1.1 Objective of the Meeting

The objective of the DMD Awareness Meeting is to reveal current problem areas based on the experiences of patients and families in the field of Duchenne Muscular Dystrophy, identify common needs, and evaluate proposed solutions to these needs multidimensionally.

1.2 Meeting Agenda

The DMD Awareness Meeting began with opening remarks following brief introductions of the participants. In the opening session, the institutional structure of the Health Institutes of Türkiye (TÜSEB) and TAÇESE was introduced by the President of the Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE), Prof. Dr. Şirin Güven; the Institute's mission, vision, and fundamental working approach were shared. Additionally, a general briefing was provided regarding the current workshops and projects carried out by TAÇESE.

Following the opening remarks, Deputy Chief Physician of Şehit Prof. Dr. İlhan Varank Training and Research Hospital, Dr. Yakup Süleymanoğlu, shared his evaluations regarding the process both as a health administrator and as a relative of a DMD patient. Subsequently, a presentation regarding the medical aspect of DMD and its current situation in Türkiye was delivered by Spec. Dr. Gülcan Akyüz Yücel from the Pediatric Neurology Clinic of the same hospital.

In the main section of the meeting, a session focusing on patient and family experiences was conducted under the moderation of TAÇESE; within this scope, problems experienced during the DMD process, unmet needs, and proposed solutions were discussed. Difficulties encountered in accessing health services, care and rehabilitation processes, and daily life were addressed in line with the views shared by the participants.

The program continued with an interactive discussion section where questions, answers, and additional recommendations were received, and the written contributions of online participants were also recorded for evaluation. The meeting was concluded with a general evaluation and closing session, running for approximately two and a half hours without a break.

The detailed agenda of the meeting is presented below.

TİME	SESSİON	SPEAKER / RESPONSİBLE
09.30 – 10.00	Opening Remarks	Dr. Yakup Süleymanoğlu, Deputy Chief Physician, Şehit Prof. Dr. İlhan Varank Training and Research Hospital Prof. Dr. Şirin Güven, President of the Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE)
10.00 – 10.10	National Perspective: Current Debates on DMD Disease in Türkiye	Spec. Dr. Gülcan Akyüz Yücel, Pediatric Neurology Clinic, Şehit Prof. Dr. İlhan Varank Training and Research Hospital
10.10 – 11.00	Patient and Family Experiences: Identification of Problems-Needs and Receiving Recommendations	Moderated by TAÇESE
11.00 – 11.30	Views and Additional Recommendations	DMD Association Representatives and Participants
11.30 – 12.00	Evaluation and Closing	TAÇESE

Table 1. Meeting Agenda

1.3 Meeting Participants

The DMD Awareness Meeting, coordinated by TAÇESE at the TÜSEB Koşuyolu campus, was held with a total of 70 participants (10 in-person, 60 online). In addition to the TAÇESE team, the representative from the Provincial Directorate of Health Public Health Services, representatives from the Türkiye Institute of Health Policies (TÜSPE), patient and family associations operating in the DMD field, as well as patients' relatives invited through these associations, researchers, and field experts attended the meeting.

2. Duchenne Muscular Dystrophy (DMD): Current Medical Framework

SPEAKER **Spec. Dr. Gülcan Akyüz Yücel** - Şehit Prof. Dr. İlhan Varank TRH Pediatric Neurology

General Information and Epidemiology

Duchenne Muscular Dystrophy (DMD) is an X-linked recessively inherited, genetically originated, and progressive muscle disease. It develops due to the deficiency or absence of the dystrophin protein in muscle tissue. While it is commonly seen in boys, clinical findings may also manifest in approximately 8% of female carriers. It occurs in one out of every 3,500–5,000 live births globally, with an estimated total patient number of around 300,000, and an estimated 5,000 patients in Türkiye.

Clinical Findings and Diagnosis

The disease is generally diagnosed around the age of 3.5–4; initial findings are seen as delays in motor development milestones, walking impairment, difficulty in climbing stairs, and toe walking or ataxic gait. Difficulty rising from a seated position (Gowers' sign) and pseudo-hypertrophy in the calf muscles are among the notable findings. Speech delay can also be observed. Elevated levels of creatine kinase (CK) is an important criterion in diagnosis, and CK values in these children are generally well above normal. Today, muscle biopsy has been replaced by genetic testing. Loss of ambulation develops around the age of 10–12 on average, and patients begin to use wheelchairs.

Complications

DMD affects not only the muscles; it also impacts the heart (cardiomyopathy), respiratory system (restrictive lung disease), skeletal system (scoliosis, osteoporosis), and neurocognitive development. The average life expectancy is considered to be around 30 years. However, with new treatment approaches, it is becoming possible to extend this period and allow patients to live independently for a longer time. Therefore, it is critical for patients to be regularly monitored by many disciplines such as neurology, cardiology, pulmonology, orthopedics, endocrinology, gastroenterology, physical therapy and rehabilitation, psychiatry, and psychology.

Current Treatment Approaches

Corticosteroids: The fundamental treatment used for the last 20–30 years. By suppressing inflammation, it slows muscle weakness, prolongs ambulation time, supports respiratory and cardiac functions, and delays the development of scoliosis. However, with long-term use, side effects such as growth retardation, weight gain, hypertension, glucose metabolism disorders, and osteoporosis are frequently observed.

Vamorolone: A next-generation steroid notable for its lesser side effect profile. It has been approved by the FDA and EMA for use in children over 4 years of age.

Givinostat: A high-cost drug that can be used in patients aged 6 and older. When administered with corticosteroid treatment, it has been shown to reduce muscle loss and inflammation, and prolong walking and standing time. It has not yet received EMA approval.

Exon Skipping Treatments: Aimed at synthesizing a smaller but functional dystrophin protein by skipping damaged gene regions. This method works with a mechanism similar to the Nusinersen (Spinraza) treatment used in SMA. It can transform DMD into the milder form, Becker Muscular Dystrophy (BMD). However, it can only be applied in patients with specific mutations.

Ataluren (Translarna): Used in children with non-sense mutations. Since dystrophin synthesis stops early in these mutations, functional protein cannot be produced. Ataluren aims to increase dystrophin production by allowing the completion of protein synthesis. However, this drug lacks FDA approval, and its EMA approval was not renewed in 2023.

Gene Therapies: Aimed at transferring genes into cells via adeno-associated viruses to ensure microdystrophin production in the body. The first gene therapy, which received FDA approval in 2023, has begun to be administered. However, the treatment is costly, and due to the large structure of the dystrophin molecule, technical difficulties and side effects

may arise during packaging and administration. This treatment is planned to be applied in patients aged 4 and older. It is contraindicated in some exon deletions.

Elevidys: One of the gene therapy options that has come to the forefront in recent years and has begun to be applied in certain patient groups. Its efficacy and safety are being closely monitored.

Conclusion

DMD is an inherited, progressive muscle disease. Although there is no definitive cure, current treatments slow the progression of the disease and improve the quality of life. While developments in next-generation steroids, targeted therapies, and gene therapy are promising, the multidisciplinary approach still remains the fundamental element in improving the lifespan and quality of patients.

Figure 1. World Duchenne Awareness Day Visuals

3. Current Situation and Needs Analysis in DMD from the Participant's Perspective

This section includes the views and determinations of the meeting participants, realized with the involvement of individuals diagnosed with DMD, caregiving families, academicians, and health administrators.

Physicians can observe the difficulties DMD patients experience in accessing health services during their clinical practice. However, the multidimensional difficulties families face in social life outside the hospital and in home care processes can only be fully comprehended through the perspective of the patient and the patient's relatives.

It is a critical need to develop and strengthen the muscular dystrophy centers currently serving under the **Ministry of Health** in terms of **multidisciplinary** operation, infrastructure, and human resources.

It is aimed that the findings and outputs obtained from this meeting will form a fundamental data set for a comprehensive and multidisciplinary workshop planned to be organized in the coming period with the participation of the relevant department presidency of the Ministry of Health, specialist physicians, and academicians.

Listening firsthand to the needs from the perspective of DMD patients and their relatives in this meeting will contribute to increasing the quality of the project, program, and policy recommendations to be developed regarding the subject.

> Importance is attached to the development of beneficial domestic and national products for our country with **TÜSEB** support. In order to reduce foreign dependency in disease management and facilitate access to treatment, the development of **TÜSEB**-supported domestic and national health technologies (drugs, medical devices, etc.) is considered a strategic priority.

> During the diagnosis stage of the disease, in cases where current genetic analyses fail to yield definitive results or remain insufficient, invasive procedures such as muscle biopsy may still be required.

> It was expressed that highly competent experts lead clinical studies directed at the DMD disease in our country. It is observed that patient relatives conduct intensive advocacy activities during the processes of importing specific innovative drugs used in patients with

single gene mutations (e.g., Ataluren) into Türkiye and including them under **SSI coverage**. Clinical observations indicate that such drug treatments can halt the progression of symptoms for several years in the early stages; however, due to the progressive nature of the disease, patients typically transition to wheelchair use around the ages of 9-10. Additionally, since the relevant specific drugs are not covered for non-ambulatory patients, steroid treatment coupled with regular physiotherapy continue to be the most critical elements in disease management at this stage. Since physical limitations and continuous sitting positions can lead to secondary mental issues like agitation and depression in patients, psychological support must absolutely be integrated into treatment protocols.

Although there are initiatives to establish multidisciplinary muscular dystrophy centers in our country by examining good practice examples in Europe and the USA, various administrative and structural obstacles are observed in the sustainability of these structures. In some examples, situating these centers in hospital annex buildings causes disruptions in specialist physician assignments and coordination problems. Furthermore, the relatively low patient volume specific to rare diseases can create disadvantages for physicians within the current supplementary payment (performance) systems. This situation negatively impacts service continuity and the motivation of professionals. For a multidisciplinary service to function effectively, these centers must be structured within the main buildings of large health complexes, such as City Hospitals, where diagnostic and imaging (EMG, MRI, etc.) infrastructure is strong. Considering that branch specialists such as cardiologists, pulmonologists, or orthopedists might lack sufficient experience regarding DMD in their routine practices; it would be beneficial to bring together experienced and willing experts in specific centers to create an institutional memory (tradition) rather than dispersing them across different institutions. Accordingly, to overcome current organizational inadequacies in university and state hospitals, it is primarily recommended to establish a **"Pilot Muscular Dystrophy Reference Center"** in Istanbul composed of experts with a multidisciplinary approach.

In addition to creating multidisciplinary centers that will provide high-quality healthcare and strengthening the existing ones, the domestic production of drugs and medical devices in rare disease management is a strategic imperative. In particular, R&D activities aimed at developing vital equipment, such as domestic ventilators, must be supported within the scope of national health policies. Considering the long-term nature of drug and medical device development processes, it is important to persist resolutely—without losing hope—in scientific studies in this field. In this context, multi-stakeholder collaboration models, formed with the voluntary participation of physicians with clinical experience, researchers, and patients' relatives who directly experience the field, bear great significance in domestic production and health policy efforts to be spearheaded by TÜSEB.

Beyond medical care processes, there is a need for holistic support mechanisms that must be provided within the scope of the social state concept. When good practice examples in developed countries are examined, it is seen that the physical spaces where the patient lives are state-supported and reorganized in a way that considers both the patient's independence and the caregiver's ergonomics. For example, in some European countries, it is observed that modifications addressing the needs of the patient and caregiver (such as installing a rail system to move the child, systems to ease bathing and toilet needs, and appropriate beds) can be provided under state services through home visits.

The fully dependent physical condition of advanced-stage DMD patients transforms hospital access and waiting periods into an unsustainable logistical and physical burden for both the patient and the caregivers. To reduce the social isolation of patients and caregiver burnout (e.g., the necessity for 24-hour uninterrupted physical support), moving health services into the home environment is a critical need. In this context, muscular dystrophy centers must work in full integration with **home health** units. Home physiotherapy services, which were successfully implemented in some institutions in the past, were halted due to deficiencies and abuses in inspection mechanisms; it would be beneficial to re-implement these services with a strict monitoring and inspection system. A comprehensive and effective home health management model is recommended to be designed in a multidisciplinary manner to include uninterrupted physiotherapy, ventilator setup and management, dietitian support, psychological counseling, preparation of medical board reports at home, medication supply, and specialist physician examinations when necessary.

The fact that disabled vehicle exemptions under current legal regulations do not cover large-volume (minibus type) vehicles suitable for wheelchair use, and the additional costs brought about by mandatory in-vehicle modifications such as ramps, pose a serious financial burden on caregivers.

Medical supplies continuously needed in disease management, such as orthoses and foot bandages, are also among the main items increasing the economic burden. Given the difficulties of transferring patients with severe mobility limitations to the hospital; the integration of measurement, fitting, and procurement processes of these customized medical products into state-supported home health services, and carrying them out through home visits by technical personnel, stands out as a structural necessity.

Before non-governmental organizations and patient representatives; emphasizing the time pressure created on families by the progressive nature of the disease, it was stated that the top priority agenda item is national and international drug development studies. The introduction of innovative drugs to Türkiye and the rapid transition of existing R&D projects, currently at the laboratory stage nationwide, into clinical application (phase trials) are

demanded. Moreover, it has been determined that there is an intense need for families to obtain information about domestic drug development projects conducted with public support (TÜSEB, etc.). While individual posts by researchers on digital platforms are an important tracking element for patients' relatives; a need emerges for such critical scientific developments to be regularly conveyed to the public through official, institutional, and transparent channels.

Unlike specific **gene therapies** that halt neuronal damage when applied in the neonatal period in the treatment of neuromuscular diseases like SMA (Spinal Muscular Atrophy); it has been determined that scientific debates regarding the efficacy of early intervention in DMD continue on a global scale. Current international literature and clinical practices focus on methods aimed at preserving the well-being (functional capacity) of patients rather than a definitive cure.

Parallel to the pursuits in the international arena, there are domestic **mRNA-based gene therapy** (dystrophin production) projects ongoing with public support in our country. In these studies, it is stated that in experiments conducted on patient-donated cells (in vitro) and transgenic animal models (in vivo), the mRNA molecule, stabilized by a new technology, provides dystrophin production independent of the mutation type (deletion, etc.). However, despite its potential in laboratory and animal experiments, clinical trials on patients (humans) (Phase 1, Phase 2, etc.) have not yet begun. Consequently, the definitive efficacy and safety of the drug on humans have not yet progressed to the verification stage. The transition from laboratory and animal experiments to human clinical trials inherently dictates exceptionally meticulous risk assessments. The progression of the process to the human trials phase depends on research protocols fully complying with national and international clinical research standards centering on patient safety. In this phase, relevant financial and regulatory procedures must be completed to prove the safety of a new drug; and demands are being voiced by researchers to accelerate and facilitate these processes without compromising scientific standards.

The management of orthopedic complications such as scoliosis that develop due to a continuous sitting position observed in the advanced stages of the disease, along with the disruptions experienced in the procurement of enteral nutrition (formula) products and medical board reports, have been identified as areas awaiting urgent systemic solutions from the perspective of patients and families.

It is observed that there are numerous non-governmental organizations and individual initiatives operating in the DMD field. It was emphasized that these structures must unite around a common vision and collaboration in order to achieve more effective results in policy development processes. In this context, with the participation of public authorities, academia, specialist physician associations, and patient NGOs; it would be beneficial to create applicable

and inclusive health policies based entirely on scientific evidence. It is considered that the obtained field data and meeting outputs will form a basis for future strategic action plans.

In light of the experiences of non-governmental organizations (DMD patient and family associations) and data obtained from patient families, the current shortcomings regarding the management of DMD and policy recommendations are identified through the following points:

- It has been identified that DMD affects not only patients but also the mothers who are genetic carriers of the disease, causing clinical pictures such as joint pains, walking difficulties, and cardiac symptoms in them. Considering the progressive nature of the disease extending from childhood to adolescence and the severe physical exhaustion experienced by caregiving families (especially mothers); it is evaluated that the topic should be one of the direct focal points of the Maternity, Child and Adolescent Health working areas.
- Making the initial support provided to gene therapy studies conducted on a national scale sustainable carries critical importance. Domestic drug development moves have strategic benefits that increase national capacity. Acting on the principle that "time is muscle," which is valid in progressive cases like DMD; it is an imperative to urgently accelerate processes in domestic research, development, and approval mechanisms aimed at rare/orphan diseases.
- Despite the existence of functional centers capable of multidisciplinary evaluation in a single day in provinces like Izmir and Antalya, the lack of such a center in Istanbul, which hosts the densest patient population, is a serious systemic deficiency. It has been determined that muscular dystrophy centers cannot be operated sustainably due to administrative difficulties such as a shortage of specialist physicians and an inability to ensure service continuity. This inefficiency reduces families' trust and application rates to existing centers, forcing patients into intercity treatment migration.
- There are hundreds of different muscular diseases in Türkiye apart from DMD, and the centers to be established must have a comprehensive infrastructure capable of responding not just to a specific disease, but to the entire spectrum of muscular diseases. It is anticipated that a functional and fully-fledged center to be established in Istanbul will be highly preferred by families, meeting the need both within the province and in surrounding provinces.
- Taking into account the physical difficulties experienced in accessing hospitals, taking measures to minimize waiting times for patients and adapting outpatient waiting areas for individuals with mobility limitations will significantly facilitate service acquisition.
- There is no uniformity of application across hospitals in the "status report signed by 3 physicians" processes mandated for physiotherapy and home education services. This

inconsistency in reporting standards causes patients with mobility limitations to wander from hospital to hospital.

- Although Ministry directives state that high percentage **Special Needs Reports** should be issued for progressive muscular diseases and their durations should be at least two years; severe grievances based on duration and percentage are experienced in practice. The lowering of disability percentages, especially after crossing the age of 18, directly cuts off adult patients' access to social services and legal rights.
- Anesthesia and surgical procedures (e.g., circumcision) carry specific risks in rare diseases like DMD. Delays are experienced in patients' access to basic surgical services due to the reservations of physicians in units unfamiliar with the disease. Functional reference centers knowledgeable on the subject will eliminate these operational risks.
- Implementing **tele-consultation** systems (digital examination) that allow remote preliminary evaluation by experts in reference centers is recommended as an important efficiency tool to prevent unnecessary referrals and patient fatigue.
- The chaos in the procurement and reimbursement processes of vital medical devices places a massive bureaucratic burden on families. In current operations, families have to shuttle between SSI depots, medical firms, and hospitals; and deal with long logistical processes like finding the device, transporting it to the hospital, and getting it approved. This situation is an unsustainable material and moral loss, especially for families whose child is in intensive care or whose condition is worsening.
- As applied in good practice examples in developed countries, periodically evaluating the process in the home environment through assigned public officials (device, catheter, physiotherapy requirement, etc.) after diagnosis, without leaving it to the initiative of the family, is a requirement of modern health management.
- It is possible to resolve the experienced disruptions with simple administrative arrangements. With a model similar to the pharmacy prescription system in drug procurement, when the need for a device is determined by experts, the equipment must be delivered directly to the patient's home/intensive care unit. In addition, managing the periodic maintenance, calibration, and home health follow-ups of the devices directly by professionals will increase the patients' quality of life.
- The early introduction of **cough assist devices** used to preserve respiratory capacity in adolescent and adult DMD patients into the system allows heavy and permanent interventions like tracheostomy to be deferred to later ages. The fact that these vital devices, whose costs currently reach the level of 170,000 TL, are not under SSI coverage prevents a large portion of patients from accessing treatment. Customs taxes and medical firm intermediary costs generated by the import-based system deepen this grievance.
- There is a precedent where domestic ventilators were successfully produced and introduced to the field in collaboration with national industry (e.g., Vestel) during the COVID-19 pandemic. Based on this precedent, it is essential to initiate the domestic R&D and production of high-cost imported devices used in the DMD field with state incentives, and simultaneously to urgently bring existing devices under SSI coverage. This investment

will not only increase the children's quality of life but also prevent the massive health costs arising from early intensive care admissions.

- Establishing a functional DMD reference center in Istanbul, supporting domestic drug/device production, and simplifying bureaucratic processes are highlighted as priority areas.

Manufacturing medical devices, procured at high costs from abroad, with domestic capabilities is considered a strategic priority in terms of health policies. In this context, to guide domestic R&D projects that can be supported via public funds (TÜSEB, etc.), turning concrete device needs obtained from patient and physician field experiences into projects bears great importance.

One of the biggest hurdles encountered in administrative processes is the inconsistencies in disabled medical board reports. Despite the progressive and irreversible clinical course of the disease, the dramatic reduction of disability percentages during report renewal periods (e.g., lowering from 90% to 60%) and their subsequent correction via appeals to referee hospitals demonstrate a lack of standardization in current evaluation criteria. Preventing such grievances, duplicate applications, and inter-institutional disputes is possible through the establishment of large-scale multidisciplinary centers that gather all relevant branches such as neurology, cardiology, pulmonology, orthopedics, PTR, and endocrinology under the same roof. These reference centers to be established will increase service quality by ensuring that single-physician, three-physician, and board reports are issued in a standard and consistent manner appropriate to the nature of the disease through competent boards.

Structural problems faced by DMD patients across many areas from education to health have been identified by the families of DMD patients:

- In the educational dimension; despite the positive discrimination and accessibility provisions in the **Ministry of National Education** legislation, planning the classrooms of students with mobility limitations on upper floors in schools and the inability to provide the necessary attendance flexibility at the higher education level increase the risk of patients dropping out of education.
- In the context of preventive health policies; the fact that 60-70% of DMD cases in Türkiye recur despite having a family history (multiple cases seen in the same family in some specific regions) clearly reveals the inadequacy of premarital genetic screening programs and public awareness campaigns (public service announcements, etc.).
- On the other hand, covering only standard wheelchairs under SSI rather than high-quality wheelchairs that protect the patient's posture accelerates secondary orthopedic

complications like scoliosis in DMD patients; similarly, difficulties in accessing vital cough assist devices continue.

- In the ethical and socioeconomic dimension of the process, the risks created by unmonitored charity campaigns are noteworthy. Directing families to treatments whose scientific validity is unproven offered in various countries (e.g., Dubai) and turning children into objects of fundraising via the media are evaluated as a serious ethical violation and area of exploitation.
- Furthermore, it has been determined that the decisions of international authorities (**FDA**, etc.) regarding drugs with debatable efficacy harbor the risk of turning our country into a market target for specific pharmaceutical companies.
- To eliminate all these risks; safeguarding the right to education, making medical device reimbursements inclusive, preventing unscientific false hope peddling, and rapidly integrating and implementing domestic gene therapy projects with ready clinical teams in the field have been determined as the core solution strategies. It is recommended that the public, academia, and civil society take joint responsibility to establish a continuously operating coordination platform.

As a result of the meeting outputs and field evaluations; it has been determined that the meeting of patients, physicians, and decision-makers on the same platform holds critical value in terms of identifying and resolving problems. It was emphasized that not only situation assessment should be done, but the obtained data must urgently be transformed into concrete and actionable action plans. In this context, under the leadership of institutions like TÜSEB, which have both academic and ministerial backing; the establishment of multidisciplinary reference centers, the domestic production of medical devices (cough assist devices, etc.), and innovative gene therapies have been designated as priority project areas. Additionally, integrating Turkish scientists working on gene therapies abroad into national projects is seen as a strategic opportunity. Alongside accelerating scientific processes and clinical research approvals; the expansion of premarital genetic screening programs, increasing public awareness through public service announcements, and providing psychosocial support not only to patients but also to caregiving mothers and families have been evaluated as the fundamental pillars of the new health policies to be established.

3.1 Written Views of Online Participants

In this section, the views and suggestions written as comments by participants who attended the meeting online have been compiled and summarized.

Emphasis on Gratitude and Contribution:

Participants expressed their gratitude for organizing the meeting and emphasized that it is especially important for the voices of families to be heard. It was stated that this meeting would create a "butterfly effect," contributing to broader awareness and a resolution process.

Education-Related Problems:

Some families mentioned experiencing problems in the admission of children with DMD into schools; for example, kindergarten enrollment was denied, and school administrations did not want to take the risk. It was reported that difficulties in access are experienced due to assigning the children's classrooms to upper floors, and there are cases that could not be resolved despite reporting this situation to the Ministry of National Education.

"Doctor, my child is not being admitted to school because he is a DMD patient. I want information regarding this to be submitted to the Ministry of National Education as well."

Access to Health Services and Report Issues:

Families stated that the obligation to renew reports every year is incompatible with the nature of the disease and puts families in a difficult position.

"Currently, the issue of the status report signed by 3 physicians must be resolved urgently."

In addition, it was expressed that up-to-date and equipped muscular dystrophy centers are insufficient, and families are forced to make intercity trips for checkups.

"We don't have a hospital where these children can have their daily tests done; we go from Istanbul to Izmir for the day. We families also experience a lot of difficulties in this regard. If only something could be done about this. Unfortunately, not every neurology doctor knows this disease. Getting every test done and making appointments is very exhausting for our children."

Some families voiced that psychological support services should also be provided at home.

"My younger brother is 19 years old right now; he cannot walk, and his psychology is very broken, and he is extremely aggressive."

"I don't think anything will improve; in this state, they make our children and their families go from door to door every year to hospitals to get reports. This is a progressive, non-regressing disease; a solution must first be found for this. And our children need to be visited at home and given psychological support."

Medical Devices and Drugs:

It was emphasized that cough assist devices, appropriate wheelchairs, and other medical devices should be provided with state support. It was voiced that accessing these devices is difficult, and families suffer grievances due to high costs. Additionally, expectations for supporting domestic drug studies were conveyed.

Some participants stated that the expectation of a "definitive cure" in the short term is unrealistic, and according to information obtained from pharmaceutical companies, a few years are needed for new treatments. Some expressed the view that there is currently no "definitive cure."

Genetic Screening, Prevention, and Counseling:

Many participants emphasized that premarital genetic screening should be made mandatory. It was stated that this could prevent the birth of multiple children with DMD in the same family.

"Reports, psychological support, and premarital genetic screening!"

A participant noted that there are significant differences among physicians in genetic screening and fertility counseling practices; while some physicians direct family members to screening and inform them about subsequent pregnancies, others do not share this information at all.

"Our doctor; directed me to genetic screening because I am his older sister, and also informed my mother not to give birth to another child. For another acquaintance of mine, however, the doctor did not provide this information in any way."

False Hope Peddling and Unscientific Methods:

Participants stated that families are exposed to false hope peddling by some associations or structures through methods lacking scientific validity, such as stem cell applications in India. It was voiced that such applications must be prevented and families should be raised awareness.

Importance of Research and Domestic Studies:

It was emphasized that domestic drug studies should be supported by TÜSEB, and rapidly implementing domestic gene therapy studies would improve the quality of life for numerous children. Expressions of hope were shared that a treatment emerging from Türkiye could be groundbreaking on a global level.

"Within the framework of the Rare Diseases Health Strategy Document and Action Plan published by the Ministry of Health in 2023, supporting clinical trials in the fastest way possible by applying the 'exceptional circumstances evaluation' will enable many children to hold on to life."

4. Expectations and Policy Recommendations Based on the Experiences of DMD Patients and Families

In light of the experiences and expert evaluations shared at the meeting, the current problems faced by individuals diagnosed with DMD and their families, and the actionable policy recommendations directed at solving these problems, are structured under the following headings:

Multidisciplinary Muscular Dystrophy Centers

- Establishing centers in university and city hospitals that bring together pediatric neurology, cardiology, physical therapy, psychiatry, dietitians, and social workers.
- Addressing the need for a functional, multidisciplinary muscular dystrophy reference center, especially in Istanbul.
- Providing DMD-specific diagnosis, treatment, follow-up, and rehabilitation services holistically in these centers, as well as facilitating reporting processes.

Home Care and Telehealth Services

- Strengthening coordination with home health services.
- Implementing home physiotherapy services, of which there were past application examples.
- Providing psychological support, ventilator procurement, orthosis measurements at home, and delivering technical team support to families.
- Reducing unnecessary referrals and travels through telehealth services.

Facilitating Health Report Processes

- Regulating the annual report renewal obligation, which creates a serious burden for families.
- Accepting long-term reports due to the chronic and progressive course.
- Eliminating inter-institutional inconsistencies in three-physician signed reports and disability report renewals.
- Developing facilitating practices that do not increase the burden on families during the report issuance process.
- Making regulations regarding discrepancies in disability percentages in health reports after the age of 18.

Psychosocial Support Programs

- Providing regular psychological support, especially for adolescent children with DMD.
- Establishing psychological and social support mechanisms for families.
- Creating support programs that will alleviate the mental burden of caregivers.

Right to Education and Social Participation

- Not rejecting the school enrollment of children with DMD and ensuring equal opportunities in education.
- Providing accessible classrooms in accordance with legislation.
- Ensuring access to activities that will improve children's quality of life.

Access to Medical Devices

- Providing SSI reimbursement for medical devices/products (Cough assist device, supported wheelchair, pressure relief cushion/bed, medical bath/toilet aids, transfer lift, communication support devices, etc.).
- Resolving ventilator/filter supply problems.
- Implementing a "medical device prescription model" that will ensure the needs assessment for devices to be used at home (respiratory support device, orthosis, etc.) and their direct delivery to the home.
- Enabling technical service and calibration to be done at home.

Domestic R&D and Drug Development Strategies

- Supporting domestic drug research.
- Allocating special funds for drugs and products targeting rare diseases, and accelerating evaluation-funding-licensing processes.
- Incentivizing domestic production and technology transfer.

Preventive Health Policies and Genetic Counseling

- Providing genetic screening programs and family counseling.
- Giving priority, standardized genetic counseling to those with a family history.
- Conducting national information campaigns (public service announcements, etc.) to prevent information pollution in society and create evidence-based awareness.

Protection Against Unethical Practices

- Protecting families against applications lacking scientific validity and characterized as false hope peddling (Stem cell treatments abroad without scientific basis and charity campaigns, etc.).

- Strengthening inspection and information mechanisms under the coordination of the Ministry of Health.

5. Evaluation and Conclusion

The DMD Awareness Meeting served as an important platform where patients and families could directly express their experiences and gather at the same table with academicians, physicians, and public authorities. The views emerging from the meeting once again demonstrated that DMD is not solely a medical problem, but also a multifaceted domain involving education, social life, psychosocial support, access to the health system, and ethical dimensions.

What the participants conveyed revealed that early diagnosis, regular follow-up, a multidisciplinary approach, and accessible support mechanisms are of vital importance due to the progressive and irreversible nature of the DMD disease. Facilitating health report processes, strengthening home care and rehabilitation, bringing medical devices under SSI coverage, and expanding genetic screening in the premarital period are among the prominent demands.

A common will was also voiced at the meeting towards supporting domestic treatment studies, accelerating research funds, and protecting families against unethical practices lacking scientific basis. It was emphasized that barriers to educational access and social participation must be removed, and children must be supported not only with health services but also with social rights.

In conclusion, the views and suggestions voiced in this meeting hold significant reference value in making the priority needs of children with DMD and their families visible. The common expectation of the participants is the transformation of these suggestions into concrete steps, thereby strengthening health services and social support mechanisms. It is anticipated that this prepared report will contribute as a guiding document for relevant institutions and decision-makers and form a strong foundation for future policy development processes.

APPENDIX

Notes shared by DMD Türkiye - Association for Combating Duchenne Muscular Dystrophy:

SSI Coverage and Procurement Model of Medical Devices Used for DMD Patients

The medical devices that Duchenne Muscular Dystrophy (DMD) patients need to use throughout their lives can vary depending on the course of the disease and the patient's needs. The fundamental medical devices generally used by DMD patients are:

1. Respiratory Support Devices

In DMD patients, advancing muscle weakness can affect respiratory functions. These devices provide respiratory support:

Cough Assist Device: This device, which is of vital importance for clearing the airways, is not under SSI coverage. Although there have been opinions for many years that it will be included in SSI coverage, it has not been covered yet. This device, whose importance was understood during the pandemic period, must be brought under SSI coverage as soon as possible.

Additionally, the devices we use in the advanced stages are:

- Non-invasive Mechanical Ventilation (NIV) Devices: Used during night sleep.
- Tracheostomy Ventilator Device: For continuous respiratory support in the advanced stage.
- Oxygen Concentrator and Pulse Oximeter: Used for monitoring and supporting oxygen saturation.

**These devices are covered by SSI, but the reimbursement rates are very low; in cases of need, if the SSI does not have them in stock (which is a very common situation), the family has to procure it themselves, pay the fee in cash upfront, and later retrieve a small portion of the cost from SSI.*

Moreover, difficulties are experienced during the procurement of equipment such as catheters and filters used with these devices. Because fewer items are provided than the patient's daily need, the family tries to cover the gap with their own means, placing an additional financial burden on the family.

2. Orthoses and Mobility Aids

Devices that facilitate mobility due to muscle weakness:

Walking Orthoses (KAFO, AFO): Used to support walking in the early stage. The SSI reimbursement rate is very low; today, while an AFO has reached 15 thousand TL, the reimbursement is around 1000 TL.

Wheelchairs:

When children stop walking around the age of 10, they are obliged to use a powered wheelchair because the children's arm muscles weaken, making them unable to be independent with manual wheelchairs. A report is required for a powered wheelchair; the board issuing the report denies the report using the children's age as an excuse, whereas our children use powered wheelchairs very well right from the start. Even if the report is obtained, the allowance provided under SSI coverage is very small. While powered wheelchairs are around 30 thousand, the reimbursement has remained around 2500 TL.

Transfer Lift (Hospital Type Lift): Used to facilitate the transfer of a large or heavy patient from bed to chair or other areas.

These lifts are not covered by SSI in any way.

3. Other Supportive Devices Not Covered by SSI:

- Pressure Relief Cushions and Beds: To prevent pressure sores.
- Medical Bath and Toilet Aids: To facilitate daily living activities.
- Communication Support Devices: Alternative communication devices for patients experiencing speech difficulties.

And the effective use and regular maintenance of these devices increase the quality of life of DMD patients. Furthermore, the devices must be regularly evaluated by health professionals.

Overview of similar health policies implemented in other countries.

In European Union countries, the products needed by DMD patients and similar patients are assessed on-site and delivered to their homes in amounts meeting their monthly needs. This prevents the family from worrying about this issue and experiencing financial anxiety.

Additionally, before patients whose respiratory tracts have been opened via Tracheostomy are discharged, the family should be given training on patient care and the use of the devices.

4. Our Suggestion for a Procurement Method Like the Pharmacy Model

All these medical products should be procured through a system at least similar to the pharmacy system, allowing families to obtain the devices by paying only their respective copayment amounts. Used devices in the hands of the SSI frequently break down, leaving our patients to suffer grievances. Also, during the repair period, a temporary device for the patient to use cannot be found; the patient is sometimes forced to stay in the intensive care unit until the device is repaired.

Current model: Procuring medical devices from private firms also causes an increase in the cost of the devices. Procuring the devices through a widespread and supervised system like a pharmacy will also lower these costs and pave the way for easier access to the devices.

In addition, domestic manufacturers should be incentivized and supported in the production of these medical devices.

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